

# Hematopoietic Stem Cell Transplant for Osteopetrosis (TCIRG1) Mutation Using Thiotepa Treosulfan Fludarabine Conditioning: Brief Report

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## Abstract

Osteopetrosis (OP) is rare metabolic bone disease characterized by impaired osteoclastic bone resorption. It is inherited by autosomal dominant or recessive mode, with variable penetrance, resulting in variable severity. It can present with cytopenia's, extramedullary hematopoiesis and neurological features like nerve compression. Hematopoietic stem cell transplant (HSCT) is the curative treatment for severe cases (Malignant Infantile Osteopetrosis- MIOT variant) however literature to support the same is very sparse. We report 3 patients with OP, two of which were successfully treated with HSCT using TTF (Thiotepa Treosulfan Fludarabine) conditioning. HSCT is curative in these patients with better outcome when done early.

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Abbreviations:

OP	Osteopetrosis
HSCT	Hematopoietic stem cell transplant
MIOT	Malignant infantile osteopetrosis
TTF	Thiotepa Treosulfan Fludarabine
DSA SAB	Donor specific antibody single antigen bead
PBSC	Peripheral blood stem cells
GvHD	Graft versus host disease

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